

# 17<sup>th</sup> Association of Sri Lankan Neurologists Annual Congress ASNAC 2024 1<sup>st</sup> – 3<sup>rd</sup> March 2024

## ORAL PRESENTATIONS

### OP 01

#### CLINICAL AND EEG CHARACTERISTICS OF CHILDREN WITH FIRST EPISODE OF SELF-LIMITED EPILEPSY WITH CENTROTEMPORAL SPIKES (SLECTS) IN A TERTIARY CARE PAEDIATRIC NEUROLOGY CENTRE IN SRI LANKA

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**Background and Objectives:** Self-Limited Epilepsy with Centro-Temporal Spikes (SeLECTS) is the commonest childhood epileptic syndrome. Behavioural and neuropsychological deficits rarely emerge or worsen during the active phase of SeLECTS although seizures usually resolve by puberty.

To describe the clinical and electroencephalographic (EEG) characteristics, behavioural and emotional problems of children presenting with the first episode of seizures to the Lady Ridgeway Hospital.

**Methods:** A retrospective cross-sectional study was carried out at three paediatric neurology units. Children with the first episode of SeLECTS were identified by screening all digital EEGs performed in 2019. Clinical details were obtained by contacting the parents. Electroencephalographic features were analysed using Nihon Kohden neurofax EEG-2100 and NicoletOne EEG systems. The child's current emotional and behavioural problems were identified using a validated version of Strength and Difficulties Questionnaire (SDQ). Data was analysed using SPSS 20.

**Results:** A total of 4756 EEGs were screened and 44 children were identified. The majority were males; mean age was 7.9 (SD=2.6) years. Mean age at first seizure was 7.9 (SD=2.6) years. Family history of

epilepsy was found in 6.8% and 9.1% gave a history of febrile seizures. Hemiclonic focal seizures (68.2%), unilateral facial sensory-motor seizures (54.5%), speech arrest (47.7%), hyper salivation (34.1%) and oro-pharyngo-laryngeal seizures (22.7%) were the key manifestations; 65% had seizures during sleep. EEG findings were typical spikes and waves in 93.2% and, 52% were in the centro-temporal region. One or more 'abnormal' scores were identified among 15 (34%) either in emotional, conduct, hyperactivity, and peer problem subscales. Ten parents (22.7%) reported having learning deficits and 32% were concerned about the impact of their epilepsy. Bilateral EEG spikes were significantly associated with high impact scores ( $p=0.008$ ).

**Conclusions:** Most children were males. Typical centro-temporal spikes and waves were identified in nearly all. Around one third of the children had some form of emotional and/or behavioural problems even at the onset of their epilepsy.

### OP 02

#### THE CLINICAL AND GENETIC SPECTRUM OF ATAXIA AND MOVEMENT DISORDERS IN A COHORT OF SRI LANKAN PATIENTS

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**Background and Objectives:** Ataxia and movement disorders are common presentations of neurogenetic diseases. Next-generation sequencing based whole exome sequencing (WES) has revolutionized the diagnosis and management of them. However, data is lacking in under-represented populations. This study describes the clinical and genetic spectrum of ataxia and movement disorders in a cohort of Sri Lankan patients.

**Methods:** Data from 28 patients presenting with ataxia and movement disorders above 14 years with suspected genetic aetiology but not confirmed with other investigations, who underwent WES between January 2015 and January 2023, was maintained prospectively and analysed retrospectively. Patients had undergone fragment analysis for Huntington disease and spinocerebellar ataxia types 1,2,3,6,7,8

prior to WES if clinically indicated and found to be negative. WES data was generated using Illumina HiSeq platform with target sequencing coverage of 100X. Copy number variations and deep intronic variations were not captured. Patients were categorised based on the predominant phenotype according to clinical presentation.

**Results:** The mean age of the cohort was 33 years. The majority - 57.1% (16/28) were males. The predominant phenotype was ataxia in 28.6% (8/28) and a movement disorder in 71.4% (20/28). Genetic variants were detected in 57.1% (16/28) comprising of 87.5% (14/16) pathogenic /likely pathogenic (P/LP) and 12.5% (2/16) variants of uncertain significance. The diagnostic yield of the cohort was 50% (14/28).

The predominant phenotypes, associated genes with P/LP variants, implicating diseases and their respective frequencies were: Ataxia 28.6% (8/28) – ATM (12.5%, 1/8) implicated in ataxia telangiectasia, ELOVL5 (12.5%, 1/8) implicated in spinocerebellar ataxia 38, PRNP (12.5%, 1/8) implicated in Gerstmann-Straussler Disease, VPS13D (12.5%, 1/8) implicated in autosomal recessively inherited spinocerebellar ataxia 4; Tremor 25% (7/28) - ATP7B (42.9%, 3/7) implicated in Wilson Disease; Atypical Parkinsonism 17.6% (5/28) - ATP7B (20%, 1/5) implicated in Wilson Disease, SPTBN2 (20%, 1/5) implicated in autosomal recessively inherited spinocerebellar ataxia 14, PINK1 (20%, 1/5) implicated in early onset parkinsonism; Dystonia 10.7% (3/28) - TOR1A (33.3%, 1/3) implicated in torsion dystonia, PANK2 (33.3%, 1/3) implicated in pantothenate kinase-associated neurodegeneration; Chorea 10.7% (3/28) - VPS13A (33.3%, 1/3) implicated in choreoacanthocytosis; and Parkinsonism with dystonia 7.1% (2/28) - PARK7 (50%, 1/2) implicated in early onset parkinsonism. Family members with similar phenotypes were noted in 39.3% (11/28) and 10.7% (3/28) were products of consanguineous marriages.

**Conclusions:** The diagnostic yield of WES was 50% emphasizing its utility in genetic confirmation of hereditary ataxia and movement disorders with implications for treatment and prognostication.

### OP 03

#### FEASIBILITY STUDY: USING BRAIN-COMPUTER INTERFACES IN STROKE REHABILITATION

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**Background and Objectives:** Physiotherapy is a major component in the rehabilitation of stroke survivors. Functional Electrical Stimulation (FES) is used as a component of physiotherapy. A Brain-computer interface (BCI) interprets the user's intent into actions. The objective was to explore the feasibility of implementing a BCI in subjects with upper extremity weakness due to stroke. We hypothesized that BCI augmented FES will enhance recovery of stroke by enhancing plasticity.

**Methods:** Six subjects were recruited, while results from three subjects who completed the study are reported. BCI+FES system was developed with the power of different EEG frequency bands and using support vector machine and random forest algorithms, with accuracies of  $52\% \pm 5.2$ ,  $54\% \pm 7.5$  and  $51\% \pm 3.9$  for each subject. For all three subjects the following functional tests were done pre-BCI and post-BCI: Fugl-Meyer Assessment (FMA), Barthel Index (BI) and Chedoke Arm and Hand Activity Inventory (CAHAI). For subjects P2 and P3, electromyography (EMG) was recorded from the extensor compartment of the forearm, while for subject P3, electroencephalography (EEG) was also recorded.

**Results:** All three subjects showed improvement in the functional scores. The total FMA score improved from  $72.67 \pm 21.502$  to  $94.0 \pm 31.607$ . The BI score improved from  $68.33 \pm 14.434$  to  $76.7 \pm 20.207$  while the CAHAI score improved from  $13.33 \pm 9.292$  to  $20.3 \pm 13.796$ . Subject P3 showed most improvement (36%, 18% and 50% improvement for FMA, BI and CAHAI respectively) while subject P2 showed the least improvement (24%, 8% and 43% improvement for FMA, BI and CAHAI respectively). EMG showed 133% improvement in P2 and 15% in P3. EEG power in mu and beta bands showed improvement of 0.5% and 0.9%.

**Conclusions:** In this feasibility study we found that BCI improved electrophysiological changes of acute stroke more than chronic stroke and it is a feasible add-on option to conventional physiotherapy. The data shows promising results as an exploratory study on the feasibility of implementing BCI for stroke rehabilitation.

#### OP 04

### EFFICACY OF THALIDOMIDE IN THE TREATMENT OF TUBERCULOUS OPTOCHIASMATIC ARACHNOIDITIS: INSIGHTS FROM A CASE SERIES

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**Background:** Optochiasmatic arachnoiditis (OCA) results from accumulation of tuberculous exudates or tuberculomas in the interpeduncular, suprasellar and Sylvian cisterns. Although there is no treatment with proven efficacy, immunomodulation has been utilized in its management. Thalidomide, which inhibits tumour necrosis factor-alpha (TNF-α) synthesis has been used for its anti-inflammatory potential. We report our experience of thalidomide in the treatment of OCA.

**Case Presentation:** Five patients with OCA without HIV co-infection were treated with thalidomide after multidisciplinary discussions. This included three re-treatment patients and two new patients (Table 1). At the time of thalidomide initiation, all except one patient had raised cerebrospinal fluid (CSF) protein. Two patients had grossly elevated protein, lymphocytic pleocytosis and CSF/blood sugar ratio <0.5. Considered treatment failure, the latter two patients were on second line antituberculous therapy (ATT). All patients received intravenous dexamethasone prior to thalidomide. One patient received pulsed methylprednisolone, followed by three doses of intravenous infliximab 300 mg at 0, 2 and 6 weeks, with suboptimal response.

The average time from start of ATT to thalidomide initiation was 6.6 months (range 5-11 months). The median thalidomide dose was 2.3 mg/kg/day with treatment durations ranging from 3 to 12 months. Thalidomide led to clinical improvement in two patients, without significant visual recovery. Increased meningeal enhancement, enlarging tuberculoma and perilesional oedema were noted in 4/5 patients (Figure 1). One patient continued to progress, necessitating surgical decompression. One patient died one month after starting thalidomide due to myocardial infarction in a background of known ischaemic heart disease. This patient had transaminitis as a consequence of thalidomide.

Rashes, cytopenia and neuropathy were not observed in any patients.

**Discussion:** Thalidomide may be of clinical benefit in selected patients with OCA despite paradoxical radiological worsening. Further studies are warranted to identify the optimal timing, dose, and duration of thalidomide.

#### OP 05

### EVALUATION OF THE TREATMENT GAP IN MANAGEMENT OF CONVULSIVE STATUS EPILEPTICUS, IN CHILDREN AT THE LADY RIDGEWAY HOSPITAL, SRI LANKA

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**Background and Objectives:** To evaluate the treatment gap for convulsive status epilepticus (CSE) in children admitted to the premier children's hospital in the country and identify possible aetiologies, and compare the treatment gap with that described in Low and Middle-Income Countries.

**Methods:** This cross-sectional descriptive study with an analytical component investigated the clinical characteristics, management, and deviations from protocols in children presenting with CSE to the Preliminary Care Unit (PCU) of Lady Ridgeway Hospital for Children, over a three-month period in 2023. All children aged three months to 15 years who presented with seizures fulfilling the International League against Epilepsy (ILAE) definition of established SE, were included. Data were gathered related to patient characteristics, aetiology, pre-hospital care, management at PCU, description of the status, and deviations from ILAE protocols.

**Results:** This study analysed 386 cases of seizures in children. The majority were 1-2 years old (28.0%) and male (59%). Most were simple febrile convulsions (68.7%). Pre-hospital care for the seizure was limited, with only 42.5% receiving any intervention. Among them was positioning (26.8%) and only two individuals received pre-hospital anti-seizure medications in the oral formulation. The total number of patients who experienced established status was 177 (45.9%) however in 148 of them the status aborted before arrival at the hospital. Febrile seizures were the commonest aetiology (67.5%). Thirty-nine arrived seizing in status epilepticus. A response to first-line medications was observed in 75%. ICU care was required for three patients, but only one received it immediately. Recognition of seizure onset was a major challenge, affecting 77.2% of cases.

Education level of both parents was significantly associated with poor recognition of seizure ( $p < 0.01$ ). Transport issues (38.3%), expert unavailability (9.1%), and drug availability (2.8%) were challenges hindering optimal management. Irregularities in medications/dosing and notes were found in eight cases (2.1%).

**Conclusions:** Pre-hospital care with anti-seizure medication is underutilized. Poor recognition of the onset of seizures was associated with low parental education. Despite most cases being simple febrile seizures, many were consistent with the diagnosis of established status epilepticus. Response to first-line treatments was frequently adequate. There was no treatment gap observed in hospital care during the study period.

#### OP 06 THE ROLE OF CYTOKINE-MEDIATED INFLAMMATION IN FEBRILE SEIZURES

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**Background and Objectives:** Immune mediated inflammation is believed to play a role in the pathogenesis of febrile seizures (FS). This study aims to compare the association of pro-inflammatory (IL-1beta, IL-6) vs anti-inflammatory (IL-4 and IL-10) cytokines between FS (cases) and non-seizure febrile illnesses (controls) in children aged six months to six years.

**Methods:** Cases ( $n=25$ ) and controls ( $n=25$ ) including 12 simple FS (SFS) and 13 complex FS (CFS) were recruited from the Lady Ridgeway Hospital, Colombo between January and October 2023. Serum cytokine levels were measured using Enzyme-Linked Immunosorbent Assays (ELISA).

**Results:** The levels of IL-1beta ( $64.38 \pm 8.25$  pg/ml vs.  $63.27 \pm 5.61$  pg/ml) and IL-4 ( $1.21 \pm 1.36$  pg/ml vs.  $0.52 \pm 0.45$  pg/ml) were elevated while IL-6 ( $59.73 \pm 46.89$  pg/ml vs.  $72.68 \pm 62.27$  pg/ml) and IL-10 ( $21.43 \pm 11.21$  pg/ml vs.  $23.22 \pm 13.65$  pg/ml) were decreased in cases compared to controls, although not statistically significant. The levels of pro-inflammatory cytokines were higher than anti-inflammatory cytokines in cases (IL-1beta vs. IL-4:  $64.38 \pm 8.25$  pg/ml vs.  $1.21 \pm 1.36$  pg/ml; IL-6 vs. IL-4:  $59.73 \pm 46.89$  pg/ml vs.  $1.21 \pm 1.36$  pg/ml;  $p < 0.05$ ).

Serum IL-1beta was elevated in CFS compared to SFS (IL-1beta:  $68.06 \pm 9.41$  pg/ml vs.  $60.4 \pm 4.3$  pg/ml;  $p=0.024$ ). Positive correlations were identified between IL-1beta and IL-4 in CFS ( $r=0.637$ ;  $p=0.019$ ) and IL-1beta and IL-6 ( $r=0.643$ ;  $p=0.024$ ) in SFS.

**Conclusions:** A pro-inflammatory cytokine-biased immune response may contribute to the pathogenesis of FS. A higher concentration of IL-1beta can be observed in CFS compared to SFS.

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#### OP 07 REVIEW OF CLINICAL AND WHOLE EXOME SEQUENCING RESULTS FROM PATIENTS WITH DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY WITH OR WITHOUT DRUG RESISTANCE (DEE +/- DRE) ATTENDING THE NATIONAL EPILEPSY CENTRE OF SRI LANKA.

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**Background and Objectives:** Describe socio-demographic, clinical, and molecular findings of a consecutive series of patients attending the National Epilepsy Centre with developmental and epileptic encephalopathy and investigated using whole exome sequencing (WES).

**Methods:** Fifteen consecutive children with DEE +/- DRE who were investigated using WES were identified using case records maintained by a paediatric neurologist and clinical geneticist. Demographic, clinical, and genetic data were analysed using descriptive statistics.

**Results:** The fifteen patients included eight (53%) males. The average age was  $1.81 \pm 2.84$  and  $4.20 \pm 3.07$  years respectively for disease onset and WES testing. There were four (27%) fathers and two (13%) mothers between 36-40 years at patients' birth. The patients were from ten districts. In six (40%) and seven (47%) patients respectively, parental income was between Rs. 100,000-200,000 and >Rs. 200,000. Epilepsy-protocol-MRI brain scans were normal in 10 (67%) while interictal-EEG remained abnormal in 11 (73%). Seizure types

included infantile spasms (7; 46.7%), generalized-motor seizures (4; 26.7%), and two each of focal-motor and focal-non-motor seizures. Eleven (73.3%) had global developmental delay. Antenatal problems and term deliveries by Caesarean section were reported in eight (53.3%) and 13 (87%) respectively. None had significant perinatal insults. Mean birth weight was 2.95 kg  $\pm$  0.63. In eight (50%), a diagnosis was made by WES including two SCN1A and one each of SCN2A, MECP2, NALCN, CACNA1D and PACS2 pathogenic/ likely pathogenic variants. A clinically suspected Dravet-syndrome patient tested negative. In all WES diagnosed patients, there was a positive impact on management. Six had variants of uncertain significance (VUS).

**Conclusions:** Molecular diagnosis is helpful in determining the genetic aetiology and improves patient management. In Sri Lanka, costs and limited access to familial segregation or functional studies limits delineation of VUS. This series illustrates the cost implications of genetic testing as most families able to get testing were high income earners.

#### OP 08

##### MEETING THE TIME TARGETS IN ACUTE STROKE THROMBOLYSIS- A CLINICAL SURVEY AT DISTRICT GENERAL HOSPITAL EMBILIPITIYA (DGHE)

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**Background and Objectives:** Time is brain. It is vital to adhere to standard time targets in stroke thrombolysis. According to the internationally accepted time targets, it is aimed to achieve a door-to-needle time (DNT) within 60 minutes in 75% or more and a DNT of within 45 minutes in 50% or more of acute ischaemic stroke patients treated with intravenous thrombolysis. A door-to-CT time (DTC) of 25 minutes or less is considered the best practice. A stroke thrombolysis service was established in December 2022 in DGHE following a series of targeted awareness program.

**Methods:** A printed protocol was utilized to document evaluation of every hyperacute stroke admission, triaged for thrombolysis. Demographic details, NIHSS score, and time taken at each step were documented. Data from December 2022 to November 2023 were collected.

**Results:** There were 22 patients who presented within the thrombolysis window, who underwent evaluation in the hyperacute stroke pathway. There were 15 males (68%). Eleven patients were thrombolysed after fulfilling the eligibility criteria.

Out of them, eight had moderate (NIHSS 5-15), one had moderate to severe (NIHSS 16-20) and two had severe (NIHSS 21-42) strokes. Out of the thrombolysed, the mean DTC time was 45 minutes, (SD= 46.48) with a range of 100 minutes (15 minutes to 115 minutes). The mean DNT was 84 minutes, (SD = 41.04) with a range of 113 (42 minutes to 155 minutes). Only 37.5 % were thrombolysed within 60 minutes.

**Conclusions:** DGHE did not meet the standard goals of thrombolysis time windows which is comparable to other published data within Sri Lanka. Measures should be taken to minimize these time windows. Restructuring the thrombolysis flow within the hospital and continuous thrombolysis training for health care workers would be of potential benefit.

#### OP 09

##### EFFECTS OF A MEDITATION-BASED INTERVENTION ON MOTOR MANIFESTATIONS OF PARKINSON DISEASE: A RANDOMIZED CONTROLLED CLINICAL TRIAL

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**Background and Objectives:** Parkinson disease (PD) is the most common neurodegenerative disorder that leads to an akinetic-rigid syndrome. Although dopaminergic therapy remains the most effective treatment for PD, it often fails to provide optimal symptom control. Thus, we explored the efficacy of an adjunct meditation-based intervention (MBI) on the motor manifestations of PD in a randomized controlled trial.

**Methods:** Forty-six patients with PD (H&Y 1-3) were selected using convenient sampling and randomized to an interventional group (IG) and a usual-care-alone group (UC). IG underwent eight weeks of MBI in addition to their routine treatment. Motor manifestations were assessed via three tests: Short Parkinson Evaluation Scale/Scales for Outcomes in Parkinson Disease-motor function (SPES/SCOPA-Motor), Timed-Up-and-Go (TUG) test and Tibial nerve-nerve conduction study (NCS). SPES/SCOPA-Motor tool assessed motor evaluation (ME), activities of daily living (ADL) and motor complications (MC). TUG evaluated functional mobility and NCS evaluated neural processing and transmission. All tests were performed before and after intervention. Data was

analysed using SPSS-29 software. Non-parametric tests were used to assess outcome significance.

**Results:** Twenty-three PD patients each were randomized to IG (M:F=14:9; mean (M:F=13:10; mean age=66.1, age=63.9, SD=6.6 years) and to UC (M:F=13:10; mean age=66.1, SD=6.7 years). Baseline characteristics of the two groups did not differ significantly. IG group had a significant improvement in results following the intervention. Post-intervention results between the two groups are; SPES/SCOPA-Motor [ME mean-rank: IG=12.4, UC=32 (SE=41,  $p<0.001$ ), ADL mean-rank: IG=16, UC=28.2 (SE=40.9,  $p=0.001$ ), MC mean-rank: IG=18.7, UC=25.5 (SE=39.3,  $p=0.06$ )], TUG test mean-rank: IG=18.4s, UC=25.3s (SE=40.5,  $p=0.05$ ) and NCS, conduction velocity mean-rank: right side IG=28.9m/s, UC=14.7m/s (SE=41.1,  $p<0.001$ ) and left side IG=28.2m/s, UC=15.5m/s (SE=41.1,  $p<0.001$ ) while amplitude mean-rank: right side IG=31.1mV, UC=12.5mV (SE=41.1,  $p<0.001$ ) and on left IG=31.4mV, UC=12.1mV (SE=41.1,  $p<0.001$ ).

**Conclusions:** MBI is an effective adjunct in improving the motor manifestations of PD.

## POSTER PRESENTATIONS

### PP 01

#### UNVEILING THE MITOCHONDRIAL MYSTERY: A CASE OF MELAS

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**Background:** Neurological disorders with mitochondrial inheritance, are a group of rare diagnoses which require a high index of suspicion. Among these, mitochondrial encephalopathy with lactic acidosis and stroke-like episodes (MELAS) is extremely rare and no cases have been reported from our part of the world.

**Case Presentation:** We came across a 38-year-old female patient with difficulty in walking and intermittent focal onset seizures with preserved awareness. She had been on treatment for epilepsy with multiple antiseizure medications since the age of 20 and had an episode of status epilepticus at the age of 25. Examination revealed nystagmus and other peripheral cerebellar signs. Lower limbs were spastic with up-going plantar responses bilaterally. An arterial blood gas revealed a raised lactate level of 3 mmol/l. Magnetic Resonance Imaging (MRI) scan of the brain revealed multiple cortical infarcts involving right temporal and parietal lobes and new T2 signal abnormalities involving old infarcts in the right occipital lobe and both cerebellar hemispheres. There was evidence of mineralization of the globus pallidus with cerebral atrophy. MR spectroscopy of the brain showed evidence of a lactate peak in the right signal region and in the cerebrospinal fluid of the lateral ventricles. Her cerebrospinal fluid lactate level was also elevated. Her anti-seizure medications were reviewed as sodium valproate is contraindicated in mitochondrial disorders and she was started on coenzyme Q10. Her seizures settled and she was referred for physiotherapy.

**Discussion:** Mitochondrial encephalopathy is an important differential diagnosis to consider in refractory epilepsy as well as in strokes among young patients. This case is a good example of how neuroimaging could play a pivotal role in the diagnosis of MELAS in the absence of genetic studies due to limited resources.

### PP 02

#### BRAIN IN BRAIN MALFORMATION, A RARE VARIANT OF MALFORMATION OF CORTICAL DEVELOPMENT

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**Background:** Brain-in-brain malformation (BBM) is a rare malformation of cortical development (MCD), noted in patients with Drug-Resistant Epilepsy (DRE). So far, this condition has not been recognized as a specific entity due to its rarity. Two cases (C1 and C2) with BBM are reported herein.

**Case Presentation:** Both patients started getting seizures during their early infancy (C1 – two months and C2 – six months). The first seizure semiologies in both were - left focal motor seizure to generalized tonic-clonic seizures (GTCS). The semiology has evolved to asymmetric epileptic spasms (C1), focal motor status epilepticus to focal non convulsive status epilepticus (C2) since the onset. Currently both have Drug Resistant Epilepsy (DRE), with epileptic tonic drop attacks (C1). C2 gets focal to GTCS, atonic drop attacks and atypical absence seizures. Both get daily multiple events. The family history, antenatal and perinatal histories were unremarkable. Both are delayed in development (all domains), with low IQ and issues in Strength and Difficulty analysis. Clinical examinations were unremarkable. Inter-ictal EEGs were compatible with epileptic encephalopathy with some focal changes. 3 Tesla MRI scans of the brain revealed the midline-to-left conglomerate masses of dysplastic brain tissues representing miniature brains with deeply in folded tissue representing gyral folds with grey matter lined cortical tissues. With incomplete lobar separation, the absence of a corpus callosum is in keeping with the interhemispheric variant of holoprosencephaly. Polymicrogyria with grey matter heterotopia is noted at different foci in the rest of the brain. C2 had periventricular band heterotopia at the left temporal region.

**Discussion:** Brain in brain cortical malformations are rare entities of MCD and may represent a severe form of subcortical grey matter and or neuro-glial heterotopia. However, the exact aetiopathogenesis and the classification are yet to be elucidated. We suggest multicentre data collection for further delineation of Brain in Brain Cortical malformation.

### PP 03

#### EPILEPSY IN INFANCY WITH MIGRATING FOCAL SEIZURES WITH MUTATION IN KCNB1 SUCCESSFULLY TREATED WITH TOPIRAMATE

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**Background:** Genetic factors are a major contributor to drug resistant epilepsy. KCNAB1 mutation causes epilepsy with temporal spikes but has not been reported to cause epilepsy in infancy with migrating focal seizures (EIMFS). We report a case of KCNAB1 associated EIMFS with migrating focal seizures controlled by topiramate.

**Case Presentation:** A Sri Lankan boy presented with refractory seizures since day 2 of life. The semiology was an abnormal cry followed by clonic movements. By five months, he had multiple daily seizures going up to twenty events a day and had global developmental delay. He was the first child born to nonconsanguineous parents without any antenatal or perinatal complications. His maternal grandmother was treated for epilepsy but didn't have any apparent developmental concerns. His electroencephalography (EEG) showed slowing over the left temporal region and migrating focal seizures of temporal origin. Extensive metabolic screening and imaging didn't reveal any aetiology. Whole exome sequencing showed him to have a heterozygous missense variant (c.1043C>T (p.Ala348Val) in the KCNAB1 gene, known to be associated with susceptibility to various forms of epilepsy with temporal spikes on EEG. Parental screening for a similar mutation was negative. His seizures remitted after adding topiramate at five months. He was started on early developmental intervention. Currently the child is two years and eight months and is seizure free and has achieved appropriate developmental milestones excepting expressive language skills.

**Discussion:** Here we report the first case of epilepsy in infancy with migrating focal seizures with KCNAB1 mutation and its successful treatment with topiramate.

#### PP 04

##### **A CASE OF LENNOX-GASTAUT SYNDROME (LGS) FOLLOWING AUTOIMMUNE ENCEPHALITIS RESPONDING TO THE KETOGENIC DIET**

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**Background:** Epileptic encephalopathies are a group of disorders that lead to unrelenting seizures associated with cognitive and behavioural decline. Lennox-Gastaut Syndrome (LGS) represents a specific epileptic encephalopathy, displaying diverse seizure patterns and a distinctive electroencephalogram (EEG). The ideal management of LGS following autoimmune encephalitis (AIE) remains a puzzle as it is a rare entity, and the pathophysiology is uncertain. We report a case of post-encephalitis LGS responding to a ketogenic diet.

**Case Presentation:** A five-year-old boy was diagnosed with AIE when he presented with typical EEG and clinical features without evidence of central nervous system infection. He developed super refractory status epilepticus and was managed with methylprednisolone pulses, intravenous immunoglobulins, plasmapheresis, and rituximab. He recovered with a modified Rankins score of two but was nonverbal and continued to have frequent atonic convulsions, progressive cognitive decline, and hyperactivity. At nine months post initial insult, the EEG was typical of LGS. There was no response to antiseizure drug adjustments, and methylprednisolone pulses. A ketogenic diet was initiated and after five months on the diet, the patient showed no clinical seizures, no epileptiform activity on EEG, and notable improvement in cognitive and speech ability and behaviour.

**Discussion:** The case expands the literature regarding the development of LGS following AIE and highlights the possibility of successful remission with ketogenic diet even in those unresponsive to anti-seizure medications and immunotherapy.

#### PP 05

##### **UNUSUAL MANIFESTATIONS OF JUVENILE DERMATOMYOSITIS (JDM)**

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**Background:** Juvenile dermatomyositis (JDM) is an inflammatory disorder characterised by muscle and dermatological manifestations. This report



details the case of a seven-year-old girl with an unusual presentation of JDM.

**Case Presentation:** A seven-year-old girl, who had been experiencing excessive hair growth two months, along with a history suggestive of proximal muscle weakness for one month, presented with a sudden onset of right-sided hemiplegia. Dermatological signs included Gottron's papules, nail fold capillary abnormalities and an uncommon occurrence of hirsutism. Creatine kinase level was elevated at 2397 U/L (normal range: 30 – 150 U/L). A magnetic resonance imaging (MRI) scan of the brain, along with MR angiogram, revealed an infarction in the left middle cerebral artery (MCA) territory, accompanied by vessel narrowing indicative of vasculitis. Antinuclear antibody (ANA) was >1:80, and the extractable nuclear antigen antibodies (ENA) panel was negative. Treatment involved the administration of methylprednisolone, aspirin, azathioprine, and cyclophosphamide. Three months later with the prescribed management and subsequent rehabilitation, the child showed improvement and is back to her normal activity without any further clinical events.

**Discussion:** JDM is a multisystem disorder where the pathology is a micro vasculitis. Medium / large vessel vasculitis is an uncommon but documented characteristic associated with JDM. On rare occasions, JDM patients may exhibit focal weakness warranting consideration of cerebral vasculitis in their diagnostic evaluation. Cerebral vasculitis is rare with no clear guidelines for management of this complication. Hirsutism is a rare manifestation of JDM that can act as a sentinel sign.

## PP 06

### CDKL5 DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY PATIENT PRESENTING WITH INFANTILE SPASMS

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**Background:** CDKL5 developmental and epileptic encephalopathy (DEE) is a recently described genetic DEE of infantile onset, due to the mutation in the cyclin-dependent-kinase-5 gene. With the advancement of genetic epilepsy panels, this is now a separate entity, characterized with early onset epilepsy, dysmorphism, movement disorders, hypotonia and corticovisual impairment (CVI). We describe a genetically confirmed patient who presented to us with infantile spasms with a classical clinical phenotype.

**Case Presentation:** A four-month-old baby, first born to non-consanguineous parents with normal antenatal and birth history presented to us with infantile spasms. His developmental age was less than six weeks. He had had infantile tonic seizures starting from six weeks onwards, which had gone unnoticed. His EEG was hypsarrhythmic and, he was commenced on the United Kingdom Infantile Spasms Study (UKISS) regimen of prednisolone with minimal response. There after vigabatrin, topiramate, lamotrigine and levetiracetam were prescribed, with unsatisfactory seizure control. Later, the ketogenic diet was commenced, and he responded well. He had severe developmental delay, hypotonia and CVI phase 1 as associated factors. His metabolic workup and MRI brain was normal. He was genetically confirmed to have a CDKL5 gene mutation.

**Discussion:** CDKL5 genetic DEE is a separate disease entity with unique features. Epilepsy predominates the clinical phenotype starting as very early onset infantile tonic or tonic clonic seizures, as the first stage. Infantile spasms are the second stage, followed by myoclonic, tonic, absence, and atonic seizures. Seizures are usually pharmacoresistant and suggested drugs are topiramate, lamotrigine, valproate, vigabatrin and zonisamide. Other modalities of treatment include the ketogenic diet, vagal nerve stimulation and ACTH. Upcoming interventions in clinical trials include cannabidiol and fenfluramine. Other associated features aiding diagnosis include dysmorphism, CVI, movement disorders, severe development delay and hypotonia. Timely identification of this specific genetic DEE will guide precision care, and hence good epilepsy and developmental outcomes.

## PP 07

### SCN8A MUTATION-RELATED EPILEPTIC ENCEPHALOPATHY AND MULTIPLE BONE FRACTURES

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**Background:** SCN8A associated epileptic encephalopathy is one of the treatable causes of intractable epilepsy in infancy. SCN8A mutation is associated with a spectrum of clinical phenotypes, including cognitive deficits, movement disorders, muscle atrophy, bone loss, and fractures. Severe bone loss has been previously described in a child and also demonstrated in mouse models of SCN8A mutation. The pathophysiology of SCN8A mutations related to skeletal complications is still not clearly known.

**Case Presentation:** These monozygotic diamniotic (MCDA) twin children were born to consanguineous parents without perinatal complications. They were developmentally normal until six months of age when they developed focal tonic and sequential seizures; followed by development of epileptic spasms. The clinical phenotypes of both children were the same, with tonic seizures followed by developmental regression. The focal tonic seizures did not respond to a combination of sodium valproate and clonazepam. However, treatment with phenytoin loading dose resolved the tonic seizures. The spasms were treated with oral prednisolone and seizures settled for the next five months. However, recurrence of seizures resulted in regression. At 1.5 years, they presented with spontaneous bone fractures involving bilateral femurs. Clinical examination revealed blue sclera with dental enamel hypoplasia in both twins suggesting a diagnosis of osteogenesis imperfecta. The serum calcium was low; phosphate level was elevated. ALP 247 U/L(122-470U/L), vitamin D 24 ng/mL(>20ng/mL), PTH 35pg/ml(10-55pg/ml). The skeletal survey suggested osteogenesis imperfecta. The DEXA scan revealed combined trabecular and cortical bone loss. IV bisphosphonate, calcium, and vitamin D supplements were started to stop further fractures. Whole exome sequencing in one of the twins revealed a pathogenic variant in exon 27 of the SCN8A gene as a missense mutation, denoted as C.5615G>A at the cDNA level. At last review, they remained seizure free on sodium valproate and topiramate.

**Discussion:** SCN8A gene mutation in DEE 13 is associated with epileptic encephalopathy. Skeletal complications, and systemic involvement warrant active assessment for bone fractures and osteopenia.

## PP 08

### INCIDENTAL DETECTION OF TWO CASES OF PYRIMIDINE DEGRADATION PATHWAY DISORDERS BY URINE ORGANIC ACID ANALYSIS USING GAS CHROMATOGRAPHY MASS SPECTROMETRY

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**Background:** Dihydropyrimidine dehydrogenase (DPD) and dihydropyrimidinase (DHP) deficiencies are two rare congenital disorders affecting degradation of pyrimidines, uracil and thymine, with a varied clinical spectrum from asymptomatic cases

to neurological manifestations. We report two cases of DPD and DHP, presenting with neurological manifestations, detected incidentally by urine organic acid assay using gas chromatography mass spectrometry (GC-MS) in the department of chemical pathology, Lady Ridgeway Hospital for Children. GC-MS technique for urine organic acid assay though primarily used to detect organic acidurias, can identify some polar compounds indicating pyrimidine degradation pathway defects.

## Case Presentation:

Case 1: A seven-month-old baby girl born to consanguineous parents with a low birth weight (1.76 kg, <-3SD) and microcephaly (28 cm, <- 3SD) was investigated for global developmental delay and hypertonia. MRI brain revealed generalized severe cerebral atrophy while the EEG was normal. High uracil and thymine with no dihydrouracil and dihydrothymine in the urine organic acid profile confirmed the diagnosis of DPD deficiency. Case 2: A 15-day-old boy of consanguineous parents with poor sucking and hypotonia developed frequent apnoeic attacks since the fourth day of life. His urine organic acid profile revealed uracil, thymine and dihydrouracil confirming DHP deficiency supported by a missense homozygous likely pathogenic variant in the DPYS gene.

**Discussion:** There is limited awareness about inborn errors of pyrimidine pathway defects (PPD). Urine organic acid analysis should be requested in patients with unexplained neurological presentations. GC-MS technology plays a minor role in the diagnosis of purine degradation pathway defects hence a laboratory identified as a centre for rare disease detection should be armed with equipment to detect more PPD.

## PP 09

### TETRABENAZINE RESPONSIVE MYOCLONUS DYSTONIA DUE TO DYT 11 MUTATION

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**Background:** Myoclonus-dystonia is a movement disorder that typically affects the neck, torso, and arms. Individuals with this condition experience quick, involuntary muscle jerks or twitches (myoclonus). About half of individuals with myoclonus-dystonia develop dystonia, which is involuntary tensing of various muscles that causes unusual positioning. In myoclonus-dystonia, dystonia often affects one or both hands, causing

writer's cramp, or the neck, causing the head to turn (torticollis).

**Case Presentation:** A three-year-old male child presented with abnormal jerky movements in both upper limbs and difficulty in walking for four months. He was the first child of non-consanguineous healthy parents. The antenatal history was uneventful and he was delivered by elective Caesarean section at 38 weeks of gestation with 2.5kg of birth weight, and had no perinatal or post-natal complications. He was developmentally normal up to two years and two months of age and then developed recurrent falls due to left side lower limb weakness. He then gradually developed jerky movements in the upper limbs bilaterally, affecting fine movements. Neurological examination was clinically normal. His brain imaging, electroencephalogram (EEG), nerve conduction studies and basic metabolic studies were normal. These movements responded poorly to Syndopa and clonazepam. Whole exome sequencing revealed a DYT11 mutation in the SGCE (epsilon-sarcoglycan) gene. Following this, tetrabenazine was started and the jerky movements significantly improved.

**Discussion:** This case history is important due to the clinical identification of the DYT11 mutation and guidance on effective management strategies in the context of dystonia.

#### PP 10

##### **A RARE VITAMIN-RESPONSIVE DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY DUE TO PACS 2 MUTATION**

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**Background:** PACS2 (phosphofurin acidic cluster sorting protein 2) gene variation is inherited in an autosomal dominant manner. It often begins with epilepsy within one week after birth. It is accompanied by facial abnormalities and varying degrees of developmental delay. PACS2 is involved in membrane trafficking, apoptosis and autophagy. Here we describe a case history of drug resistant epilepsy which was caused by PACS2 gene variation.

**Case Presentation:** A 12-year-old child presented with multi drug resistant epilepsy. He was the first child of nonconsanguineous healthy parents. The antenatal period was unremarkable. He was

delivered by an elective Caesarean section at 40 weeks of gestation, and had no perinatal or postnatal complications. He was well up to two months of age and then developed right sided tonic clonic convulsions lasting one minute, which were followed by post-ictal drowsiness for 45 minutes. Seizure burden was 1-2 episodes per day which was complicated with global developmental delay. EEG revealed epileptic encephalopathy and MRI did not reveal an abnormality. He was treated with multiple antiepileptics, including steroids, for 11 years but had poor response. Whole exome sequencing revealed a PACS2 gene mutation and then folinic acid was added with pyridoxine. Following this seizure burden decreased by 50%. and quality of life improved significantly according to QOLCE-55 (Quality of life in Childhood Epilepsy Questionnaire).

**Discussion:** This case history is important for clinical identification of the PACS2 gene mutation and guiding effective management strategies in the context of drug resistant epilepsy.

#### PP 11

##### **PARANEOPLASTIC MYELOPATHY: A DIAGNOSTIC DILEMMA IN LONG SEGMENT TRANSVERSE MYELITIS**

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**Background:** Non-classic manifestation of paraneoplastic myelitis is rare compared to the classic paraneoplastic neurological symptoms, and even more rarely presents as long segment transverse myelitis. The aim of presenting this case is to discuss the diagnostic dilemma that occurs due to paraneoplastic myelopathy when it presents in the early stages of a tumour.

**Case Presentation:** 41-year-old lady with diabetes for six years, presented with back pain for two weeks followed by bilateral lower limb numbness, weakness, urinary retention, and bowel involvement over one day duration. She had a history of low-grade fever over two months duration prior to the onset of symptoms. Examination revealed bilateral lower limb spastic paralysis with a positive Babinski sign and a sensory level at T10. Long segment transverse myelitis involving C6 to D6 was noted on magnetic resonance imaging (MRI) with associated right mediastinal lymphadenopathy. Other investigations were as follows: slightly elevated inflammatory markers with negative screening for autoimmune and infectious aetiologies.

Cerebrospinal fluid analysis revealed a protein of 81mg/dL and 65 lymphocytes, with normal cytology. The lymph node biopsy showed reactive nodes and the bone marrow study was normal. She was mobile with improvement in bladder and bowel function following five doses of intravenous methylprednisolone. On follow-up, two months later she complained of evening pyrexia and abdominal pain. Reimaging with contrast enhanced computed tomography (CECT) abdomen revealed enlarged previously noted lymph nodes with hepatomegaly and splenic lesions. Her erythrocyte sedimentation rate (ESR) was elevated and repeat lymph node biopsy confirmed nodular lymphocyte predominant Hodgkin's lymphoma with possible transformation into diffuse large cell lymphoma.

**Discussion:** Paraneoplastic myelitis can be overlooked during evaluation due to its rarity and the nonspecific nature of the investigation findings. This case underscores the significance of considering rare possibilities when faced with a diagnostic dilemma and emphasizes the importance of close monitoring for the emergence of new symptoms, even after initial clinical improvement.

#### PP:12

#### UNILATERAL 7TH NERVE PALSY AS THE CLINICAL PRESENTATION OF MALIGNANT HYPERTENSION: THE IMPORTANCE OF BLOOD PRESSURE MONITORING IN THE PAEDIATRIC POPULATION WITH NEUROLOGICAL SYMPTOMS

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**Background:** Lower motor neurone type 7th nerve palsy is a common clinical presentation in the paediatric population and about 80% are diagnosed with Bell's palsy with unknown aetiology. Malignant hypertension can account for up to about 8% as an aetiology, secondary to renovascular stenosis, congenital malformations of the urinary system, coarctation of the aorta or endocrinopathies. In those cases, the child might only have the 7th nerve palsy without any other symptoms or signs of secondary aetiology.

**Case Presentation:** A four-year-old was admitted with a left-sided lower motor type facial nerve palsy. On the day of admission, he developed epistaxis. The child had frequent hospital visits for recurrent episcleritis and had constitutional symptoms over a six-month duration. On examination, the child had stage 2 hypertension with a blood pressure reading of 220/180 mmHg. There was no pulse difference in

all four limbs. The cardiovascular system and eye examination were normal. The Renal Doppler study revealed renal artery stenosis in the right kidney and upper pole of the left kidney, confirmed by a computer tomography renal angiogram. Other vessel involvement was excluded through MRA, MRV and angiogram of thoracic vessels. Inflammatory markers were normal, and infections were ruled out. The diagnosis of Takayasu arteritis was made with radiological findings and managed with methotrexate, intravenous methylprednisolone pulses, and later with infliximab. Blood pressure control was achieved with four types of antihypertensives.

**Discussion:** In our patient, the key indicator for blood pressure monitoring was epistaxis, which facilitated the early diagnosis of malignant hypertension. While arterial hypertension is a known cause of lower motor 7th nerve palsy, the literature indicates a median delay in diagnosis of approximately 45 days. Another concern is that in cases of misdiagnosis, a child might be prescribed steroids as a treatment for Bell's palsy, potentially exacerbating hypertension and leading to a catastrophic event.

#### PP 13

#### POSTERIOR REVERSIBLE ENCEPHALOPATHY SYNDROME (PRES) DUE TO TWO UNUSUAL CAUSES

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**Background:** Posterior reversible encephalopathy syndrome (PRES) is a syndrome of heterogeneous aetiologies characterised by altered mental status, headache, seizures, and visual changes with white matter vasogenic oedema in the posterior cerebral hemispheres on neuroimaging. We present two cases of PRES with two unusual aetiological factors.

#### Case Presentation:

Case 1: A 42-year-old female on treatment for uterine fibroids with Gonadotrophin Releasing Hormone (GnRH) agonists (Goserelin) presented with acute onset, thunderclap headache one week after the sixth dose of GnRH agonist. On admission, she developed a generalised tonic-clonic seizure. Blood pressure was 228/119mmHg. Magnetic Resonance Imaging (MRI) of the brain showed bilateral symmetrical T2/FLAIR hyperintensity white matter changes in the occipital lobes and minimal similar small areas in the posterior parietal lobe white matter bilaterally.

Case 2: A 43-year-old female with menorrhagia and uterine fibroids presented with a haemoglobin of 5.3

g/dL. Three blood transfusions were given over three consecutive days. One and a half hours after the final transfusion, the patient developed a headache. Blood pressure was 150/100mmHg with bibasal crepitations in the lungs. The patient was managed as transfusion-associated circulatory overload with diuretics. Five days after the initial onset of the headache she developed blurring of vision, worsening headache, and three episodes of generalized tonic-clonic seizures. MRI brain showed bilateral symmetrical T2 high signal intensity involving subcortical and deep white matter of occipital, parietal & paracentral gyri indicative of PRES.

**Discussion:** GnRH agonist and blood transfusion related PRES is a very rare presentation which clinicians need to be aware of.

#### PP 14

##### ACUTE ISCHAEMIC STROKE FOLLOWING HYMENOPTERA STINGS

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**Background:** Wasps and honey bees belong to the order Hymenoptera and their stings are commonly encountered in the daily life of a Sri Lankan villager. Although allergic reactions are well documented, neurological complications such as acute ischaemic stroke (AIS) are exceptionally rare. This case series reports two distinct cases of AIS following hymenoptera stings, highlighting the potential for this severe complication and the need for further investigation into its underlying mechanisms.

##### Case Presentation:

Case 1: A 58-year-old non-smoking gentleman developed sudden right-sided hemiplegia and aphasia within one hour of receiving stings from about 100 wasps in the jungle. Imaging confirmed acute infarctions in the external border zone areas on the left side of the brain. Extensive investigations revealed no pre-existing risk factors for stroke and the patient received standard stroke therapy with rehabilitation.

Case 2: A 64-year-old tea-plucker presented with sudden onset left-sided hemiplegia and dysarthria following stinging of approximately 20 honey bees. She had angioedema of the face with possible laryngeal oedema requiring supplemental oxygen in the initial few hours following admission. A non-contrast computed tomography (NCCT) scan of her brain revealed a right-sided lenticular striate artery territory infarction. She had no prior history of

stroke or any other risk factors. She was given steroids in addition to standard stroke care.

**Discussion:** The exact mechanism behind the development of ischaemic stroke following hymenoptera stings is poorly understood. Several potential pathways have been described which include; direct vascular toxicity of venom components inducing vascular inflammation, vasoconstriction and platelet aggregation, immune-mediated endothelial dysfunction and vascular damage and venom induced dysrhythmias contributing to stroke development. This case highlights the need to increase awareness among physicians to consider the possibility of AIS in the setting of neurological deficits following a hymenoptera sting even in the absence of traditional risk factors and the importance of rapid recognition and timely intervention to improve outcomes in AIS.

#### PP 15

##### ACUTE MOTOR AND SENSORY AXONAL NEUROPATHY (AMSAN) PRESENTING WITH T8 SENSORY LEVEL – A RARE CASE REPORT

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**Background:** Guillain-Barre Syndrome (GBS) is an acute immune-mediated disorder affecting the peripheral nervous system, causing acute flaccid paralysis. While classically recognized by ascending motor weakness, GBS rarely presents atypically with profound sensory level involvement, challenging diagnostic norms, and leading to diagnostic and management delays. The Acute Motor Sensory Axonal Neuropathy (AMSAN) variant of GBS, presenting with a sensory level, is an extremely rare occurrence.

**Case Presentation:** A 63-year-old Sri Lankan female, diagnosed with hypertension and ischaemic heart disease, was admitted with progressively worsening lower limb numbness and weakness over six days. By the fifth day, she developed bilateral hand numbness and urinary retention. There was no preceding history of infection. Neurological examination revealed bilateral flaccid paraparesis, generalized areflexia, and a sensory level at T8. The rest of the neurological examination and other system examinations were normal. The nerve conduction study demonstrated intact bilateral sural nerve sensory responses with absent median and ulnar responses. There were absent common peroneal motor responses and F-wave abnormalities supporting a diagnosis of AMSAN. Cerebrospinal fluid revealed albuminocytological dissociation. Magnetic Resonance Imaging (MRI) of the whole

spine was unremarkable except for L1-L5 diffuse disc bulging causing mild canal stenosis. The patient showed a dramatic response to intravenous immunoglobulin (IVIg) and further improvement with physiotherapy, experiencing mild residual motor weakness in bilateral lower limbs.

**Discussion:** In AMSAN, as in AMAN, the pathology is consistent with immune-mediated primary axonal injury at the node of Ranvier, with the difference being that both the dorsal and ventral roots are affected. It can be hypothesized that the initial inflammation at the nerve roots up to the level of T8 could have caused the symmetric T8 sensory loss in our patient clinically. This case emphasizes the need for awareness of atypical GBS presentations to improve patient care, highlighting clinical presentation, diagnostic challenges, and treatment outcomes.

#### PP 16

##### **WHEN VISION TOOK A DIVE DUE TO A MISSION IN THE SINUSES- A CASE OF BILATERAL RHINOGENIC OPTIC NEURITIS CAUSED BY A SPHENOIDAL MUCOCELE PRESENTING AS AN ALTITUDINAL FIELD LOSS**

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**Background:** Sphenoidal mucocoeles are rare complications of chronic sinusitis, that rarely can cause vision loss through optic nerve compression. Early diagnosis and intervention are crucial to prevent permanent impairment.

**Case Presentation:** A 64-year-old man presented with progressive bilateral vision loss, severe headache, and ptosis in the right eye for two months. Examination revealed complete vision loss in the right eye and limited vision in the left. He had ophthalmoplegia and an inferior altitudinal visual field defect. Fundus examination showed optic disc pallor, and magnetic resonance imaging (MRI) confirmed bilateral sphenoidal mucocoeles compressing the optic nerves with underlying bone expansion and remodelling.

**Discussion:** The patient underwent endoscopic sinus surgery, successfully draining and removing the mucocoeles. Postoperatively, his vision improved significantly in both eyes, with headache relief and reduced optic disc pallor. Histopathological analysis confirmed chronic inflammation in the mucocoeles.

This case highlights the importance of considering sinus pathology in individuals with sudden vision loss and headache, especially with atypical presentations like altitudinal field defects. Prompt collaboration between otorhinolaryngologists and ophthalmologists is crucial for optimal management and vision preservation.

#### PP 17

##### **CONGENITAL MYASTHENIC SYNDROME (CMS) - COLQ MUTATION IN A CHILD**

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**Background:** Congenital Myasthenic Syndromes (CMS) are a genotypically and phenotypically heterogeneous group of neuromuscular disorders, due to mutations of proteins in the motor end plate. Mutations occur de novo or can be inherited. They are characterized by fatigability and weakness. The onset varies from intrauterine to adolescence. The prevalence is not known as CMS is rare compared to the acquired type. In Sri Lanka, there are few reported cases in adults. We report the first case of COLQ-CMS diagnosed at three years of age in the Sri Lankan context.

**Case Presentation:** A three-year-old girl presented with respiratory arrest, following a respiratory tract infection which required ventilatory support. Atracurium was used as a muscle relaxant. She was born to nonconsanguineous parents; but isolated gross motor delay and poor weight gain were noted at six months. Stridor and swallowing difficulty were detected for which nasogastric tube feeding and physiotherapy were started. She was able to walk and take oral feeds at two years of age. She had ptosis, external ophthalmoplegia, and unequal pupil sizes. There was central hypotonia, fatigable proximal muscle weakness, reduced deep tendon reflexes with normal gait. The spine and other systems were normal. Sustained upward gaze and single breath count to twenty demonstrated fatiguability. Electromyogram (EMG) showed a decremental pattern. Pyridostigmine worsened the weakness and was replaced by salbutamol to which she responded. Whole Exon Sequencing (WES) revealed CMS with a COLQ mutation.

**Discussion:** Twenty-two CMS disease genes have been identified to date. Mutations in the acetylcholinesterase (AChE) collagen-like subunit gene, COLQ, are the most common causes of synaptic CMS. This leads to deficient AChE. Most

patients are disabled early with respiratory difficulties and axial muscle weakness. Common triggers for relapses are infection, esterase inhibitors and puberty. DOK7 and slow channel CMS may mimic COLQ-CMS. Muscle relaxants are used cautiously as the action of AChE is unpredictable. Atracurium is used in children due to its noncumulative metabolism. Succinylcholine is contraindicated. Genetic diagnosis is essential for appropriate drug selection. Pyridostigmine worsens symptoms in COLQ CMS, as in our patient. Clinical improvement has been reported with ephedrine and/or salbutamol. Our patient responded to salbutamol with improved exercise tolerance, but external ophthalmoplegia remained with minimal disturbance to activities of daily living.

#### PP 18 SUBACUTE COMBINED DEGENERATION OF THE SPINAL CORD PRESENTING AS A GUILLAIN-BARRE SYNDROME (GBS) MIMIC

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**Background:** Subacute combined degeneration of the spinal cord (SACDS) is a disease affecting the lateral and posterior columns of the spinal cord, primarily due to demyelination. It is a neurological complication of vitamin B12 deficiency. We present a nine-year-old girl who presented with ascending motor paralysis and sensory symptoms, who responded to treatment as for GBS and diagnosed as SACDS subsequently.

**Case Presentation:** A nine-year-old apparently healthy girl presented with ascending type motor weakness over ten days. Her examination confirmed lower limb more than upper limb proximal more than distal weakness with absent reflexes. Her initial sensory examination was normal. Nerve conduction studies showed only abnormal F waves in motor recordings. She developed inability to stand without support and was treated for GBS with intravenous immunoglobulin (IVIg) and improved markedly, becoming ambulatory without assistance. Over the next five days, she rapidly deteriorated and plasmapheresis was undertaken, with no improvement. By day twenty, her clinical picture evolved, and she developed paraesthesia involving lower limbs, her gait was ataxic, and her joint position sensation became impaired. On inquiry, she was a vegan for several years and started consuming animal products a week prior to the onset of weakness. Her vitamin B12 level was 101.7pg/ml (200- 800). Her MRI spine confirmed the diagnosis of SACDS. There was no haematological involvement. We prescribed, intramuscular vitamin

B12 100micrograms weekly injections for a month and once a month thereafter with dietary management, and she recovered fully by four months.

**Discussion:** SACDS, is a rare diagnosis and its presentation can vary. This case was unusual for the acute onset of isolated motor manifestations with areflexia which is attributable to motor neuropathy at presentation and the initial response to IVIg which may point to an inflammatory accompaniment of B12 deficiency-associated neuropathy.

#### PP 19 LONGITUDINALLY EXTENSIVE TRANSVERSE MYELITIS (LETM) AS THE SOLE MANIFESTATION OF *Neisseria* *meningitides* INFECTION

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**Background:** Acute myelopathy secondary to meningococcal meningitis is considered a rare complication that is limited to a few reported cases all of which occurred in the presence of typical symptoms of meningitis and signs of meningeal irritation. In contrast, myelopathy appearing as the sole manifestation of *Neisseria* infection is extremely rare.

**Case Presentation:** A 70-year-old previously healthy male presented with fever for three days and acute onset lower limb weakness with urinary retention. On examination, he was febrile, with stable vital parameters. Neurology examination revealed flaccid paraplegia with a sensory level at T2 which progressed to C4 level within 12 hours from the onset. Magnetic resonance imaging (MRI) revealed features of longitudinally extensive transverse myelitis extending from the craniovertebral junction to the T4 spinal level. His cerebrospinal fluid (CSF) showed neutrophilic pleocytosis (2560) with an elevated protein level. Both gram stain and culture of CSF were normal, but CSF was positive for *Neisseria meningitides* antigen. He was treated with IV Ceftriaxone and a course of IV methylprednisolone followed by four cycles of plasma exchange with marked improvement. Unfortunately, he succumbed to death on day 12 of hospital admission due to hospital-acquired pneumonia.

**Discussion:** *Neisseria meningitides* is considered one of the most frequent pathogens causing bacterial meningitis which may typically present with fever,

headache, vomiting, and neck stiffness. Without timely treatment, it can result in complications, including meningococcal sepsis, obstructive hydrocephalus, or cranial nerve palsies. However acute spinal cord dysfunction secondary to *Neisseria* infection is considered rare. Most reported cases have first had meningitis then spinal cord dysfunction as a sequel. Up to now, only one case has been reported in the literature to have acute myelitis as the sole manifestation of *Neisseria* reported by Ibrahim et al. in Doha, Qatar.

## PP 20

### NEUROLEPTIC MALIGNANT SYNDROME IN ANTI-N-METHYL-D-ASPARTATE RECEPTOR ENCEPHALITIS – A CASE REPORT

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**Background:** Anti-N-methyl-D-aspartate receptor (anti-NMDAR) encephalitis is a difficult diagnosis presenting with neuropsychiatric manifestations. Neuroleptic malignant syndrome (NMS) not only shares most of its features with anti-NMDAR encephalitis, but also it can develop as a consequence of antipsychotic use in these patients.

**Case Presentation:** We describe a 30-year-old male with no previous psychiatric history, who initially presented to the Psychiatry unit (PU) with aggressive behaviour and altered level of consciousness (LOC) and was given antipsychotics. He developed one generalised tonic clonic seizure and reduced LOC and was transferred to our neurology unit. High serum creatinine phosphokinase (CPK) and creatinine were detected at the PU before the seizure. On admission, he had a high fever, generalised muscle rigidity, and hyperreflexia. He was managed at the intensive care unit with respiratory support, intravenous meropenem, and acyclovir and treated as for NMS with hydration and bromocriptine. Further history revealed that he initially had a low-grade fever and headache with behavioural abnormalities. Despite the initial management, he had persistent fever with reduced LOC and developed orofacial dyskinesia. There was an ill-defined T2 hyperintensity in the left insular cortex and external capsule with no contrast enhancement or diffusion restriction in his initial magnetic resonance imaging (MRI) of the brain which was reduced in size and hyperintensity in the repeat MRI. His cerebrospinal fluid anti-NMDAR antibodies became positive. He had a marked improvement following initial immunotherapy and is currently doing well.

**Discussion:** As patients with autoimmune encephalitis with psychiatric symptoms are initially managed with antipsychotics, they have a high chance of developing NMS. This further complicates the disease process leading to diagnostic difficulty and treatment delay. Timely identification and immunotherapy lead to better outcome. More studies are warranted to differentiate whether NMS is caused by the antipsychotics given or if it could be a feature of anti-NMDAR encephalitis.

## PP 21

### CRYPTOCOCCAL MENINGITIS IN IMMUNOCOMPETENT YOUNG ADULTS

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**Background:** Cryptococcal meningitis (CM) is a relatively rare infection with high mortality and morbidity. It is considered as an opportunistic fungal infection, classically described in immunocompromised patients. Cryptococcosis has been very rarely reported in young and otherwise healthy patients. Here we report two cases of CM in young healthy females.

#### Case Presentation:

Case 1 - A 24-year-old lady presented with fever, headache, altered behaviour for 4 days duration. On examination, she had low GCS. Initially, she was managed for possible viral encephalitis. With poor response and cerebrospinal fluid (CSF) findings suggestive of pyogenic or tuberculous (TB) meningitis, antibiotics and anti-TB regimen medications were commenced. Her initial magnetic resonance imaging (MRI) brain was normal and her repeat MRI brain showed extensive cerebral venous sinus thrombosis and bilateral infarcts. Despite ongoing treatment, she had a continuing fever with headache and a persistent CSF sugar drop. Her CSF and blood cryptococcal antigen were positive. She was treated with amphotericin followed by oral fluconazole. She improved with treatment and was investigated for an immunocompromised state, but the investigations were negative.

Case 2 - A 22-year-old lady presented with fever, headache, photophobia, and diplopia for 3 days duration. On examination, she had a left 6th cranial nerve palsy. Based on her MRI and CSF findings, she was managed for pyogenic meningitis. Later, she developed seizures with a reduced level of consciousness. Her repeat CSF study showed a persistent sugar drop. An anti-TB regimen was commenced. Despite treatment, she had a continuing fever. She also was found to have hydrocephalus for



which an external ventricular drain (EVD) was inserted. Her CSF for cryptococcal antigen was positive and she was commenced on amphotericin. Her GCS improved and her fever settled. She later had obstruction of the EVD and had multi-drug resistant (MDR) *acinetobacter* and MDR *enterococci* in the CSF as well as blood cultures. Her repeat MRI had shown basal ganglia and large middle cerebral artery (MCA) infarcts with hydrocephalus. Despite aggressive treatment for sepsis, she succumbed.

**Discussion:** The most common manifestation of cryptococcal infection in an immunocompromised patient is central nervous system (CNS) involvement. Cryptococcal meningitis in young, previously healthy patients is very uncommon. The diagnosis of CM is confirmed if the patient meets any one of the following criteria: CSF-positive culture, positive India ink or positive cryptococcal antigen. Lack of awareness among treating clinicians and low index of suspicion leads to the diagnosis often being delayed or missed, resulting in poor prognosis with severe consequences.

## PP 22

### VALIDATION OF THE PROGRESSIVE PLANNING TEST (PPT) FOR USE IN THE ASSESSMENT OF CHILDREN WITH NEURODEVELOPMENTAL DISORDERS IN SRI LANKA.

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**Background and Objectives:** The Progressive Planning Test (PPT) is a “look-ahead puzzle” developed to assess cognitive planning. The main objectives of the current research were to test the following hypotheses: 1) The test performance increases linearly with age until adolescence; 2) The effect of age varies by the difficulty level of the problems, with only the problems of higher levels (levels two and three) showing age differences; and 3) Solving level two and three problems involves dynamic interplay between motor and higher cognitive processes (frontal).

**Methods:** Participants: Sixty-seven children, 33 girls and 34 boys ranging in age from six to 17 years, participated. They were typically developing children who met the following exclusionary criteria: known neurological illness, traumatic brain injury, neurodevelopmental disorders, and significant psychiatric problems. The PPT was administered individually as part of a larger

neuropsychological test battery. In this test, a participant was asked to solve 10 planning problems of graded difficulty progressing through three levels.

**Results:** The participants were grouped into three age categories and the planning problems, into three levels. One-way ANOVA showed that average total score earned by the participants increased with age linearly,  $F(2,65)=12.24$ ,  $p<.001$ . Repeated measures factorial ANOVA revealed a significant interaction between age and level of difficulty,  $F(2,64) = 5.97$ ,  $p = 0.004$ , indicating only level two and three problems discriminated between the three age groups, confirming hypothesis two. The third hypothesis was also confirmed, indicating the influence of motor habits in cognitive planning.

**Conclusions:** The current findings on the PPT are in line with the results obtained in other countries, thus attesting to its construct validity. Therefore, the PPT can be used to assess the executive functioning of children in Sri Lanka. This test can also be used to assess adults with various neurological conditions like leukoencephalopathy and Parkinson disease.

## PP 23

### QUALITY OF CARE & LEVEL OF SATISFACTION AMONG CAREGIVERS OF CHILDREN ATTENDING PAEDIATRIC NEUROLOGY CLINICS IN A TERTIARY CARE HOSPITAL IN COLOMBO, SRI LANKA

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**Background and Objectives:** The Colombo South Teaching Hospital (CSTH) is a leading tertiary care centre with two crucial clinics - Multidisciplinary Neurodevelopmental Clinic and Paediatric Neurology Clinic. Despite limited resources and time, both clinics serve a large number of patients. Thus, it is essential to assess caregiver satisfaction for improved care quality. To evaluate the quality of care and satisfaction level of caregivers of children who attend the Multidisciplinary Neurodevelopmental Clinic and Paediatric Neurology Clinic.

**Methods:** Interviewer administered questionnaires were given to caregivers attending the above clinics at CSTH over two months.

**Results:** Fifty carers were interviewed. Most children were boys (68.8%) and aged 5-10 years & 1-2 years. The primary caregivers are mothers, who accompany children for early intervention due to prematurity, complicated neonatal period (32%), cerebral palsy (18%), and epilepsy (24%). Parents expressed high satisfaction levels during registration (65%), with supporting staff (65%), nursing staff (71%), and doctors (79%), despite waiting for 30 minutes to an hour. The study highlighted the provision of excellent services through the best counselling with good insight at 64.6% & timely referrals for evaluation of vision (84.8%,) hearing (77.3%,) swallowing assessment (40%,) and oral-hygiene (31%,). Satisfaction rates for physiotherapy (88%), speech and language therapy (80%), and occupational therapy (79%) emphasizes the top-notched care.

**Conclusions:** Based on the study, parents reported that they were satisfied with the quality of care provided by both clinics. It also highlighted the importance of extending high-quality care, including counselling, complication assessment, appropriate interventions, and effective therapies. Compassionate care and efficient therapies have played a vital role in minimizing complications, allowing children to maintain their age-appropriate abilities, thus making early intervention more successful.

#### PP 24 COMPARISON OF URINE GLYCOSAMINOGLYCAN EXCRETION BETWEEN CHILDREN WITH AUTISM SPECTRUM DISORDER AND TYPICALLY DEVELOPED CHILDREN

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**Background and Objectives:** The objective of the present study was to compare urine excretion of sulphated glycosaminoglycans (GAG) in children with autism spectrum disorder (ASD), with neurotypical controls.

**Methods:** Random urine samples were collected from children with autism spectrum disorder (n=61)

between the ages of two and six years, and age- and sex-matched neurotypical controls. Urine glycosaminoglycan levels were quantified by the dimethylmethylene blue (DMMB) dye-binding assay, using a microplate reader.

**Results:** Urine GAG levels are significantly higher ( $p = 0.026$ ) in the ASD group when covariates such as age, weight, urinary creatinine, and height are taken into consideration by ANCOVA. In neurotypical subjects, the urine glycosaminoglycan levels appear to decline with age, height, and weight while this trend was not apparent in subjects with autism spectrum disorder.

**Conclusions:** Children with ASD exhibit higher urine GAG excretion. Further research is needed to explore the molecular basis of this finding.

#### PP 25 USE AND MISUSE OF URGENT EEGS IN PAEDIATRIC PRACTICE; AN AUDIT DONE IN A TERTIARY CARE CHILD NEUROLOGY CENTRE IN SRI LANKA

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#### **Background and Objectives:**

Electroencephalography (EEG) is a non-invasive investigation done to analyse the electrical activity of the brain. This audit aimed to investigate the true necessity of urgent EEG (uEEG) in paediatric patients at a tertiary care centre in Sri Lanka. The uEEG guidelines in this centre were formulated based on the 'EEG in acute Neurology' guideline of Harvard Medical School – Massachusetts General Hospital (MGH) epilepsy services. Altered level of consciousness of different strata was taken as the prime indication to perform an uEEG in this guideline.

**Methods:** A retrospective cohort analysis was conducted on paediatric uEEG requests. uEEG request forms and reports were examined by two researchers who were blinded. The deviations from the predefined guideline were assessed.

**Results:** A total number of 812 EEGs were done during a period of 12 months, out of which 402 (49.5%) were uEEGs. The mean age for uEEG was 7.59 years ( $\pm 4.58$  years). Clinical indications ranged from analysis of the first event 154 (38.3%), altered level of consciousness 103 (25.6%), breakthrough

events 77 (19.2%), to seizure with fever 57 (14.2%). Abnormalities were detected in 122 (30.3%) of uEEGs. Focal-interictal discharges 52 (46.4%) and epileptic-encephalopathy 23 (20.5%) were the commonest abnormalities. uEEG done for breakthrough events had the highest percentage of abnormal EEGs 37/77 (48.1%). Out of the nonurgent EEGs, 107 (26.1%) had abnormal reports. There was no significant difference between the percentages of EEG abnormalities between uEEG versus nonurgent EEGs ( $p = 0.18$ ).

**Conclusions:** Nearly half of the EEGs were done on urgent EEG requests. Assessment of the first event was the commonest indication. Altered level of consciousness was the indication only in a quarter and aligned with the guideline. Positive rates between EEGs done on urgent versus non-urgent basis did not show a significant difference. This audit, highlights the value of reiterating the EEG guideline among requestees to refine the requesting methodology.

#### PP 26 USE OF SCALP EEG IN CHILDREN WITH “FEVER PLUS SEIZURES”, A SINGLE CENTRE RETROSPECTIVE COHORT STUDY

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**Background and Objectives:** The co-association of “Fever-Plus-Seizures” includes febrile seizures, central nervous system (CNS) infections and first epileptic seizure or a relapsed epileptic seizure triggered by fever. A correct diagnosis of “Fever-Plus-Seizures” leads to appropriate management. The scalp electroencephalogram (EEG) is one investigation which is being requested in this clinical context. The current study was done to determine the use of scalp EEG records in children with “Fever-Plus-Seizures”.

**Methods:** A retrospective collection of data was done, from November 2022 to December 2023. EEGs requested for “Fever-Plus-Seizures” between the ages of 1 month – 16 years were included. EEG request forms and reports were independently analysed by the researchers.

**Results:** A total number of 139 EEGs were done for “Fever-Plus-Seizures” over a period of 12-months and 97/139 (69.8%) were done on urgent requests. The mean age was 5.3 years ( $\pm 3.6$ ) and 89 (64%)

were males. The indications were febrile seizures in 119 (85.6%), out of which 49 (41.2%) had complex febrile seizures, 36 (30.2%) had simple febrile seizures and 34 (28.5%) had late-onset febrile seizures (febrile-seizures occurring after an age of 5 years). In the febrile seizure group EEG abnormalities were noted in 11 (9.2%) of which six had postictal background slowing. Eight (16.3%) patients with complex febrile seizures had EEG abnormalities, (interictal-epileptiform abnormalities = 04). One with late-onset febrile seizures had interictal-epileptiform abnormalities. Epileptic seizure triggered by fever was the indication for an EEG in 11 (7.9%), with EEG abnormalities reported in 27.2%, (postictal-slowing = 1, interictal-epileptiform-abnormalities = 2). Nine (6.5%) scalp EEGs were done for clinically suspected CNS infection and abnormalities noted in 22.2%.

**Conclusions:** In a wide majority with “Fever-Plus-Seizures” the indication for a scalp EEG was febrile seizures, and a quarter of the sample had simple febrile seizures. Only a minority of the EEGs done for febrile seizures had abnormalities. Very few of the complex febrile seizure group had interictal epileptiform abnormalities. Scalp EEGs requested for the analysis of CNS infection and for seizures triggered by fever are minimal.

#### PP 27 A STUDY ON SLEEP DISTURBANCES IN CHILDREN WITH CEREBRAL PALSY

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**Background and Objectives:** According to the evidence children-with-cerebral-palsy (CWCP) are at risk of having sleep disturbances. However, this has not been considered a sinister health issue to date. Therefore, we intended to determine the frequency, types and associations of sleep disturbances in CWCP.

**Methods:** A descriptive-cross-sectional study method was used. The Sleep Disturbance Scale-for Children (SDSC) was used as the tool, which is a 26-item parent reported questionnaire with a five-point scale (SDSC, T-score of  $>70$  was taken as pathological). A trained doctor performed the rest of the assessments using validated tools.

**Results:** The sample comprised 400 CWCP; mean age 5.9 years (+/-3.3 years), males 229 (57.25%); Quadriplegic-Cerebral-Palsy (CP) n=163 (14.75%), Diplegic-CP n=130 (32.5%), Hemiplegic-CP n=89 (22.25%). Out of the sample 181 (45.25%) had at least one pathological sleep disturbance. Disorders-of-Initiating-Maintaining-Sleep (DIMS) was the commonest category n=91 (50.27%), which was identified more in children aged 2-6 years (n=65, p=0.0105) versus > six years, those with quadriplegic-CP (n=60, p=0.0001), patients with epilepsy (n=43, p=0.0001), those with a higher Gross-Motor-Function-Classification-Scale (p<0.0001), those with a higher Manual-Ability-Classification-Scale (p<0.0001), and those with a higher Communication Function-Classification-System (p<0.0001).

Out of the cohort, n=133 were on antiepileptic medication, with higher DIMS scores (n=48, p<0.0001). Sleep-Hyperhidrosis was seen in 60 (33.15%), Sleep-Breathing-Disorders in 49 (27.07%), Disorders-of-Arousal in 31 (17.13%), Sleep Wake-Transition-Disorders in 30 (16.57%), and Disorders-of-Excessive-Somnolence in 23 (12.70%). Of the sample, 388 (97.00%) co-share the bed with a family member, and sleep has not been discussed as a medical issue in 301 (75.25%). However, only a minority 66 (16.5%) considered sleep difficulties to have a negative impact on the quality of life of the family.

**Conclusions:** Nearly half of the cohort had at least a single sleep disturbance. Disorders-of-Initiating-Maintaining-Sleep is the commonest disorder, followed by Sleep-Breathing-Disorders and Disorders-of-Arousal. Sleep disturbance has not been discussed as a medical issue in the majority.

## PP 28

### EPIDEMIOLOGY, AETIOLOGY AND VISUAL OUTCOME OF CHILDREN RECEIVING REHABILITATION SERVICES FOR CEREBRAL VISUAL IMPAIRMENT IN A TERTIARY CARE HOSPITAL

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**Background and Objectives:** Cerebral Visual Impairment (CVI) is the loss of visual function resulting from damage to structures in the posterior visual pathway (beyond the lateral geniculate body). Our centre has been carrying out rehabilitation services for children with CVI for over 12 years. The analysis of the characteristics of the population and the outcome of intervention is undertaken as this

information is essential to improved detection and service delivery.

**Methods:** A retrospective observational study was conducted to identify the epidemiology, aetiology, and visual outcomes of patients followed up at the CVI intervention clinic at the Lady Ridgeway Hospital for Children.

**Results:** Data was available for 485 patients. 56.4% of them were males. 36.1% were detected at the screening when they presented to the early intervention clinic at an age of less than six months. 65.6% were reported to have a perinatal insult with 56.1% having radiological evidence of hypoxic ischaemic encephalopathy (HIE). 50.6% had respiratory distress syndrome. 64.6% of the cohort had global developmental delay and 35.6% had an abnormal electroencephalogram (EEG). Data for improvement following interventions was available in 216. Median improvement was 39.28%. Respective improvements of 25-50%, >50-75%, and >75-100% were seen in (67) 31%, (49) 22.7% and (36) 16.6% of the cohort. The level of CVI at the first visit was associated with maturity (p value = 0.037), presence of HIE (p value = 0.037) & age at the first visit (p < 0.005). The CVI score at the first visit predicted the outcome of interventions with a sensitivity of 90% and specificity of 80% (area under the curve 0.9, 95% CI 0.93-0.98, p<0.0001).

**Conclusions:** The dominant and potentially preventable cause for CVI was HIE. Neurological co-morbidities were seen in the majority which could point to CVI association that justifies screening. The usefulness of intervention was demonstrated with an improvement of 50% or more of the CVI score in more than one-third of the population, with this number increasing to more than two-thirds for an improvement of 25% or more.

## PP 29

### A DETAILED ANALYSIS OF PARENTAL PERCEPTIONS OF AUTISM SPECTRUM DISORDER (ASD): A TERTIARY CARE EXPERIENCE IN SRI LANKA

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**Background and Objectives:** Raising a child with autism spectrum disorder (ASD) can be an overwhelming experience for parents and families as ASD is characterized by poor social communication, repetitive behaviours, and restricted interests. The

objective of this study is to identify associations of parental perceptions of children with ASD, as it is of utmost importance to provide individualized parent and child-centred therapy to achieve a better outcome.

**Methods:** A descriptive cross-sectional study was conducted among 100 children with ASD meeting DSM V criteria (aged 2-17) at Teaching Hospital Kurunegala following ethical clearance from the Sri Lanka College of Paediatricians. Data was collected through an interviewer-administered questionnaire on parental perceptions using the Likert scale.

**Results:** The population comprised a mean and median age of 6.88 and six years respectively, with a male majority (76.4%). Parental perception of aetiology, social stigma, schooling, prospects and therapy were assessed. 63.5% of parents believed it is a disorder of the brain and 84% disagreed that it is familial in aetiology. With regards to social stigma, 51% found it difficult to take their child to public places, and only 11% felt embarrassed. 76.5% and 65.3% of parents believed that their child can join school and communicate with others respectively. Only 16.5% of parents feared the child would never be able to find a job, compared to 48.3% who thought otherwise. A majority (81.1%) of parents believed that doctors can improve the child's condition, of which 75.3% thought the available therapies are beneficial, but only 51.8% believed doing therapy at home was effective.

**Conclusions:** Getting a better understanding of parental perceptions will facilitate clinicians to deliver interventions that would yield higher parent satisfaction.

### PP 30 EFFECTS OF A MEDITATION-BASED INTERVENTION ON NON-MOTOR SYMPTOMS OF PARKINSON'S DISEASE: A RANDOMISED CONTROLLED CLINICAL TRIAL

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**Background and Objectives:** Parkinson's disease (PD) is characterized by motor and non-motor symptoms (NMS). There is no disease-modifying therapy yet available for PD. Currently available symptomatic therapies often fail to optimally control NMS. Thus, we explored the efficacy of an adjunct meditation-based intervention (MBI) on the NMS of PD in a randomized controlled trial.

**Methods:** Forty-six patients with PD (H&Y 1-3) were selected on convenient sampling and randomized to an interventional group (IG) and a usual-care-alone group (UC). IG underwent eight weeks of MBI in addition to their routine treatment. NMS were assessed in the two groups pre- and post-intervention using the SENS-PD (SEverity of Nondopaminergic Symptoms in Parkinson's Disease) scale. The SENS-PD assessed cognitive functioning (CF), psychotic symptoms (PSY), postural instability and gait difficulty (PIGD), excessive daytime sleepiness (EDS), autonomic dysfunction (ANS), and depressive symptoms (DEP). Data was analysed using SPSS-29 software. Non-parametric tests were used to assess outcome significance.

**Results:** In the IG there were 14 men and nine women (mean age of 63.9; SD=6.6) years. In the UC group, there were 13 men and 10 women (mean age of 66.1, SD=6.7) years. The baseline characteristics of the two groups did not differ significantly. In IG, SENS-PD scores showed a significant improvement post-intervention in all assessed parameters except psychotic features [CF mean-rank IG=16.1, UC=28.2, (SE=40.8, p=0.002), PSY mean-rank IG=22.1, UC=21.9, (SE=39.7, p=0.98), PIGD mean rank IG=14.6, UC=29.8, (SE=40.4, p<0.001), EDS mean-rank IG=18, UC=26.2, (SE=38.8, p=0.024), ANSD mean-rank IG=16.1, UC=28.2, (SE=40.2, p=0.001), DEP mean-rank IG=17.4, UC=26.8, (SE=40.6, p=0.013)].

**Conclusions:** MBI is an effective adjunct in improving the NMS of PD.

### PP 31 PREVALENCE OF PARKINSON'S DISEASE SYMPTOMS AND ITS EFFECT ON PATIENTS IN A DEVELOPING SOUTH ASIAN COUNTRY - A PATIENT'S PERSPECTIVE

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**Background and Objectives:** Parkinson's disease is a neurodegenerative condition with a range of causes and clinical presentations. Though the initial observations were limited to motor manifestations,

it became apparent that there are a whole array of non-motor symptoms, which significantly affect the quality of life of Parkinson's patients, affecting individuals to varying degrees. This study aims to describe the prevalence of the motor and non-motor symptoms of Parkinson disease and how the patients' quality of life (QOL) and activities of daily living (ADL) are being affected.

**Methods:** This is a descriptive cross-sectional study with an interviewer-administered questionnaire for patients diagnosed with Parkinson's disease according to the UK Brain Bank criteria attending the neurology clinic at the National Hospital of Sri Lanka for one year duration. Patients diagnosed with major psychiatric illness were excluded. Patients were asked to rank the impact of symptoms on their quality of life.

**Results:** Of the 192 patients, the majority were males (58.3%; n=112). The mean age of the population was  $64.09 \pm 8.69$  years. 92.7% (n=178) were married and 78.6% (n=151) had secondary education. 82.3% (n=158) rated that Parkinson's disease had an impact on employment and earning capacity. The mean age of onset of Parkinson's was  $59.5 \pm 9.87$  years. The mean duration of Parkinson's disease was  $4.62 \pm 4.25$  years. Of 192 patients, 97% (n=186) reported at least two non-motor symptoms, with fatigue (88%; n=169) and pain (74%; n=142) being identified as the most bothersome. About 83% (n=160) screened positive for anxiety and 40% (n=76) for depression. Daytime sleepiness was present in 68.7% (n=132) and insomnia in 65.1% (n=125).

**Conclusions:** Our findings provide evidence for the diversity of experience with both motor and non-motor symptoms affecting the patients' quality of life. This highlights the importance of managing both motor and non-motor symptoms to preserve the quality of life of patients with Parkinson's disease.

## PP 32

### A NOVEL GAME PLATFORM FOR IMPROVEMENT OF HAND FUNCTIONS IN STROKE REHABILITATION

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**Background and Objectives:** Stroke is a debilitating condition. Traditional rehabilitation is often costly and geographically limited, prompting exploration into novel stroke rehabilitation methods. The objective is to create a game platform using motion tracking, tailored to individual patient needs and track the progress of the rehabilitation.

**Methods:** Here we integrated motion tracking technology, employing OpenCV and Python to capture and process patient hand movements. Hand gestures were integrated for control of three off-the-shelf games. These were transmitted to the Unity game engine, enabling the generation of realistic avatar movements mapped to the hand gestures. Visual feedback was enabled and different levels of difficulty used to track progression.

**Results:** Three distinct games were developed. Game 1 "rock pick up" requires the patient to sweep rocks on the screen using their palms. The difficulty is increased by placing rocks closer in proximity. Game 1 targets palm and wrist improvement across three escalating difficulty levels. Game 2 "running man" focuses on enhancing fingers and wrist movements. Improvements of each of the fingers in both hands can be analysed. Once the finger digit is selected for playing, the game ignores other digits and only focuses on the specified digit. The difficulty can be changed by changing the speed of the avatar. In Game 3 "roll a ball", patients should virtually push the ball using the fist to collect gold and can stop the ball by squeezing the hand in a grip to avoid obstacles. Difficulty is increased by adding limitations for the game movements like adding more corners and moving obstacles. A secure login system connected to a local database grants exclusive access to patients, ensuring the therapy records.

**Conclusions:** This game platform, an innovative methodology leveraging motion tracking and personalised game experiences to enhance therapy for stroke patients shows great potential to use for rehabilitation.

## PP 33

### PREDICTING LIKELIHOOD OF IDIOPATHIC INTRACRANIAL HYPERTENSION FROM IMAGING: A RETROSPECTIVE STUDY

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**Background and Objectives:** Idiopathic intracranial hypertension (IIH) is diagnosed by the modified Dandy criteria. Magnetic resonance imaging (MRI) is used to exclude secondary causes. The clinician's dilemma is 'to LP (Lumbar puncture) or not to LP' in an individual patient, when the MRI is reported as consistent with IIH in patients who have undergone neuroimaging for a headache syndrome without clinical features of IIH. This retrospective study was carried out with the aim of identifying which MRI features are statistically significant in patients with suspected IIH.

**Methods:** MRI images of all patients diagnosed with IIH according to modified Dandy criteria and an age and gender-matched group of patients who had a diagnosis of migraine were re-reviewed by a neuroradiologist who was blinded to the diagnosis and clinical history.

**Results:** When each of the MRI features were considered separately (univariate analysis), seven features were statistically significantly associated with IIH ( $p < 0.05$ ). However, after adjusting for multiple comparisons and excluding collinearity, only two features (optic nerve sheath distension, and right Meckel's cave anteroposterior diameter) were associated with a diagnosis of IIH (Bonferroni adjusted  $p$  value  $< 0.005$ ). However, none of these features were independently associated with IIH when combined in a logistic regression. Thus, should not be used singly.

**Conclusions:** We agree that patients who are reported by radiologists as likely IIH need further evaluation. While no individual feature could predict IIH, a combination of features had a good sensitivity, specificity, positive and negative likelihood ratios. Imaging features identified in this study as being associated with IIH may be potentially useful to train an artificial intelligence-based algorithm to predict the likelihood of IIH from MRI, which in turn may be independent of the experience of the interpreter.

#### PP 34

##### UTILIZATION OF TB GENEXPERT AND TB CULTURE IN CEREBROSPINAL FLUID IN THE NATIONAL HOSPITAL OF SRI LANKA: A CLINICAL AUDIT

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**Background and Objectives:** To audit the process of cerebrospinal fluid (CSF) testing for tuberculosis (TB): CSF sampling, GeneXpert, TB culture.

**Methods:** A prospective clinical audit was carried out in 102 patients. The process of testing was analysed. Focus was on strength of ground of suspecting TB- availability of baseline chest X-ray, at least a non-contrast computed tomography (NCCT) brain, CSF full reports, and the adequacy of sampled CSF volume.

**Results:** 96 patients were treated as a possible central nervous system (CNS) infection including viral, bacterial, fungal, or TB. 13 patients were on anti-tuberculosis treatment (ATT). Out of them, seven were for central nervous system tuberculosis (CNS TB), two for TB spine (TBS), and four for pulmonary TB (PTB). TB was eventually microbiologically confirmed in one PTB and one TBS patient. All the patients except one, had undergone an NCCT before the lumbar puncture. 80% had been screened for PTB with a chest X-ray. Despite the availability of the CSF full report within 24 hours, TB GeneXpert and culture were sent before seeing the CSF full report. All the samples requested TB GeneXpert to be performed. 78 samples proceeded to be used for TB culture. 24% of samples were  $< 0.5$ ml. 74% had  $\geq 0.5$ -2ml (TB GeneXpert was positive in two). TB GeneXpert was negative in all four TB culture-positive patients.

**Conclusions:** The inadequacy of CSF sample volume is highlighted. Inefficient utilization of TB GeneXpert and culture is apparent - i.e. not considering TB culture testing in patients who are likely to benefit and on the other hand, requesting the test for patients without strong clinical grounds. Reference material on CSF sampling, for treating physicians and ground-level staff would be beneficial to fill this gap in communication. It will help to cost-effectively utilise the technical and human resources.

#### PP 35

##### A MULTICENTRE STUDY ON THE IMPACT OF EPILEPSY ON SCHOOLING IN CHILDREN

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**Background and Objectives:** In Sri Lanka, education is compulsory for children of the age group 5-16 years. Any child who leaves school before completing 16 years is considered a school-dropout. Epilepsy is the most common chronic neurological condition in children and neurocognitive and psychosocial problems associated with epilepsy may have an impact on schooling. Therefore, this study was aimed to determine the characteristics of school dropouts and to explore the factors leading to dropping out of school or never attending school in children with epilepsy (CWE).

**Methods:** A descriptive cross-sectional cohort study method was used. The study was conducted in three Teaching Hospitals of Sri Lanka. Children with epilepsy between 5-16 years were included.

**Results:** The sample comprised 207 CWE. The mean age was  $11.4 \pm 3.1$  years, males 121 (58.5%). 129 (62.3%) had focal seizures, 78 (37.7%) had generalised seizures and 52 (25.1%) had epileptic encephalopathy. Three (1.4%) children have never attended school and 34 (16.7%) dropped out. The mean age of dropping out was  $11.1 \pm 2.5$  years. 167 (81.9%) were enrolled in mainstream school, 20 (9.8%) in special school and 17 (8.3%) in special education units of a mainstream school. Of those who were attending school, 98 (57.6%) had > 75% (> 15/20 days) school attendance, 38 (22.4%) had 50 – 75% and 34 (20%) had < 50% days of attendance. Disabilities of the child was a reason for dropping out in 33 (97.1%), mismatch/non-acceptance by the school in 18 (53.3%), and financial issues in 8 (23.5%). Dropping out rates were significantly higher with higher Global Assessment of the Severity of Epilepsy (GASE) scale scores ( $p < 0.05$ ), and had no significant relationship with the monthly family income.

**Conclusions:** Only a minority have never attended school, and the majority of children were attending a main stream school. Dropping out was positively associated with severity of epilepsy.

## **PP 36 PRELIMINARY ANALYSIS OF STUDY OF LONG-TERM OUTCOMES OF A COHORT OF PATIENTS THROMBOLYSED FOLLOWING ACUTE ISCHAEMIC STROKE AT A SINGLE TERTIARY CARE CENTRE IN SRI LANKA**

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**Background and Objectives:** Data on post-thrombolysis outcomes of Sri Lankan stroke patients is limited. The study aimed to describe the National Institutes of Health Stroke Scale (NIHSS), modified Rankin Scale (mRS) and Barthel Index (BI) scores at 30 and at 90 days post-thrombolysis, and to explore the potential factors affecting them.

**Methods:** This prospective study from July to October 2023, enrolled consecutive patients thrombolysed according to standard protocols, at our neurology unit.

**Results:** Data of 37 patients (males – 24, 64.8%; age –  $61.6 \text{ years} \pm 22$ ) was analysed. Time from symptom onset to reperfusion was  $209 \text{ min} \pm 101.6$ . Only 13 (35.1%) were thrombolysed within 3 hours, with the rest between 3 and 4.5 hours. The majority (94.5%) received rTPA. Major complications included fatal ICH (2), non-fatal symptomatic ICH (5) and severe COPD exacerbation (1). 13 (35.1%) at admission, 27 (72.9%) at 30 days and 31 (83.7%) at 90 days had a NIHSS of five or below. 11 (29.7%) at 30 days, and 13 (35.1%) at 90 days were functionally independent (mRS of 0-2). 27 (72.9%) at 30 days, and 29 (78.3%) at 90 days were considered to have moderately/mildly severe disability (BI over 60). Two tailed Wilcoxon signed rank test proved statistically significant ( $p < 0.05$ ) improvements in median values of NIHSS at admission, at 30 days and at 90 days ( $Z = -3.93, -2.84$ ), mRS at admission, at 30 days and at 90 days ( $Z = -4.05, -2.66$ ), BI at admission, at 30 days and at 90 days ( $Z = -3.91, -2.51$ ). Those who had a Large Vessel Occlusion (LVO) had higher odds (OR=25.6,  $p < 0.01$ ) of being functionally dependent (mRS >2) at 90 days, whereas being male, having diabetes, hypertension, heart disease, atrial fibrillation, prior stroke or TIA, poorer social support, or presence of major complications did not have significant odds of having mRS >2 at 90 days.

**Conclusions:** The studied cohort demonstrated improvements in NIHSS, MRS and BI at 30 days compared to admission values, and at 90 days compared to 30-day values. Of the studied variables, the presence of LVO had higher odds of being functionally dependent at 90 days.



**PP 37****IMPULSE CONTROL DISORDERS AND OTHER NON-MOTOR SYMPTOMS IN SRI LANKAN PATIENTS WITH PARKINSON'S DISEASE**

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**Background and Objectives:** The impact of non-motor symptoms is often overlooked in favour of the motor symptoms when managing Parkinson's disease, resulting in suboptimal patient outcomes. This study aimed to characterise the impulse control disorders and other compulsive behaviours (ICDs-CB) of Parkinson's patients in Sri Lanka.

**Methods:** All patients with idiopathic Parkinson's disease followed up at the National Hospital of Colombo, Sri Lanka were included. An interviewer-administered questionnaire was used. Symptoms of anxiety and depression were assessed with the Hamilton Anxiety and Depression scales. Presence of ICDs-CB was assessed with the questionnaire for Impulsive-Compulsive Disorders in Parkinson's Disease.

**Results:** Of 192 patients, only one patient (0.5%) reported being aware of having a compulsive behaviour, while all the others had never been screened for or informed about ICDs-CB. According to QUIP results, 32 patients (16.7%, 32/192) screened positive for at least one ICDs-CB. The subcategories were as follows: compulsive gambling (1.6%, 3/192), compulsive sexual behaviour (1%, 2/192), compulsive buying (4.2%, 8/192), compulsive eating (8.3%, 16/192), hobbyism (6.3%, 12/192), punding (1%, 2/192), walkabout (10.9%, 21/192), compulsive medication use (0.5%, 1/192). Among these 32 patients, 15 (46.9%) had two or more ICDs-CB (two ICDs-CB: four patients; three ICDs-CB: five patients; four ICDs-CB: five patients; five ICDs-CB: one patient). A lower Barthel index, history of past psychiatric disorders and family history of alcohol abuse were independent predictors of ICDs-CB in adjusted analysis by logistic regression.

**Conclusions:** Managing both motor and non-motor symptoms are important to preserve the quality of life of patients with Parkinson's disease. They should be screened for symptoms of anxiety and depression

regularly during follow-up and educated about the possibility of ICDs-CB soon after diagnosis.

**PP 38****IMPACT ON THE RANGE OF MOTION (ROM) OF THE SHOULDER JOINT FOLLOWING HEMIPLEGIC SHOULDER PAIN (HSP) AND ITS RELATIONSHIP BETWEEN DOMINANT SIDE AND NON-DOMINANT SIDE AMONG INDIVIDUALS WITH STROKE IN GAMPAHA DISTRICT**

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**Background and Objectives:** This study was conducted to assess the impact on range of motion (ROM) following hemiplegic shoulder pain (HSP) and its relationship between the dominant and non-dominant sides in individuals following a stroke in the Gampaha District.

**Methods:** A descriptive cross-sectional study was carried out within stroke survivors with HSP (n=263) in District General Hospital, Gampaha, Colombo North Teaching Hospital and Rehabilitation Hospital, Ragama, Sri Lanka. The socio-demographic data were collected by an interviewer administered questionnaire. Pain intensity, disability level and range of motion of the affected upper limb were measured using the Numerical Pain Scale, Disability of Arm, Shoulder and Hand (DASH) questionnaire and goniometer respectively.

**Results:** Prevalence of HSP which occurred within seven days after the onset of the stroke was 29.3% (n=77). Comparing the mean of limited active ROMs (AROMs), the most affected AROM of the shoulder joint were shoulder flexion (89.35±67.800) and external rotation (42.13±32.170). Comparing the mean of limited passive ROMs (PROMs), the most affected PROMs of the shoulder joint were flexion (155.00±29.450), abduction (147.39±31.620) and external rotation (71.41±19.920). The AROM and PROM of the affected shoulder had a significant negative correlation with the aggravated pain severity. AROM and PROM in the affected shoulder were significantly associated with chronicity of stroke and also with DASH score categorization (p<0.05). Only the PROM in flexion, abduction and external rotation were significantly associated with the dominance of the affected side (p<0.05).

**Conclusions:** According to its relationship with the dominant side and non-dominant side among individuals with stroke in the Gampaha District; the PROM in flexion, abduction and external rotation were significantly affected among participants with the non-dominant side affected compared to those with the dominant side affected, but the AROM was not significantly different between these two groups.

#### PP 39

#### CLINICAL PROFILE AND OUTCOMES OF CEREBRAL VENOUS SINUS THROMBOSIS IN A TERTIARY CARE SETTING IN SRI LANKA

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**Background and Objectives:** Cerebral venous sinus thrombosis (CVST) is a rare form of stroke affecting the young, with a broad aetiology. This study aims to assess the demographic and clinical profile, as well as the management and outcome and to determine the associated risk factors for thrombosis in CVST patients in Sri Lanka.

**Methods:** This is a prospective observational follow up study for patients with a radiologically confirmed diagnosis of Cerebral Venous Sinus Thrombosis. Demographic, clinical and management related details were collected and patients were followed up during a 6-to-12-month duration.

**Results:** Of the 28 patients, the majority (82.1%, n=23) were females with the median age of the population being 35 years. The commonest symptoms were headache at 89.3% (n=25), seizures and focal neurological deficits (42.9%, n=12 each). The commonest risk factors were oral contraceptive use in 25% (n= 7), local infections in 14.7% and anti-phospholipid syndrome (APLS) in 10.7%. Radiological features consisted of sinus hyperdensity in 28.6% (n=8), cerebral oedema and empty delta sign in 14.3% each (n=4). 42.9% (n=12) had a single venous sinus involved. The commonest sinuses to be involved were the superior sagittal and transverse sinuses in 53.6% (n=15) each. Venous infarcts and haemorrhages accounted for 78.6 % (n=14). Isolated intracranial hypertension was detected in 32.1% (n=9), visual loss in 10.7% (n=3), and recurrent CVST and arterio-venous fistulas in 7.1% (n=2) each. All patients received anticoagulation. The majority (96.4%,n=27) recovered. During follow up magnetic resonance

scanning, partial recanalisation of sinuses was evident in 21.4% (n=6).

**Conclusions:** The Sri Lankan profile of CVST is mostly similar to regional and international studies, although there is a high rate of associated local infections and a hypercoagulable state. Pregnancy as an associated factor was lower in number. An increased frequency of isolated intracranial hypertension was detected. Comparatively, long-term visual complications were high. The majority had a good outcome from CVST.